Diffuse Venous Malformation with Intraosseous Involvement

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A young boy with a large venous malformation of his left arm and hand was evaluated for Maffucci's syndrome. Initial hand films were remarkable for cystic bony lesions suggestive of enchondromas. Additional roentgenograms and magnetic resonance imaging studies showed the bony abnormalities were the result of a venous malformation infiltrating bone. Magnetic resonance imaging is a useful diagnostic tool in differentiating enchondromas from vascular anomalies. Venous malformations infiltrating bone should be included in the differential diagnosis of Maffucci's syndrome.

Skeletal angiomas comprise less than 1% of all bony lesions and predominantly afflict women during the fifth decade of life.[1-3] There is a predilection for involvement of the vertebra and skull (75%).[4] Angiomas of long bones occur less than 10% of the time and have a higher association with soft tissue cavernous component than other sites.[1-4] Although the roentgenographic findings of angiomas in the skull, spine and flat bones are often characteristic, involvement of long bones may lack these features and present a diagnostic challenge.[5] This is particularly true for the congenital syndromes such as Klippel-Trenaunay-Weber syndrome and Maffucci's syndrome where skeletal angiomatosis have been reported to be associated with both conditions.[1,6] This is an account of a diffuse venous malformation with intraosseous involvement, which on clinical examination and initial hand films was suggestive of early Maffucci's syndrome.

Introduction

A 13-year-old Caucasian male came to the dermatology clinic for information on congenital hemangiomas. Shortly after birth the patient developed matted telangiectasias involving the left shoulder and arm. During his childhood years the sites evolved into large subcutaneous masses extending from the left shoulder to the tip of his thumb. Therapeutic attempts at wrapping the arm were unsuccessful. More recently he noted increased frequency and severity of his limb pain. In addition, the patient experienced a midshaft radial fracture and distal ulnar fracture with minimal trauma to the left arm.

The physical examination revealed the left arm had large bluish subcutaneous masses extending from the biceps region to the tip of the thumb (Fig 1). The left shoulder had patches of matted blue varicosities and telangiectasias. There was no limb length discrepancy or evidence of other sites of cutaneous involvement.

Fig 1.—Vascular anomaly extending from the left shoulder to thumb.

Bilateral hand films demonstrated radiolucent defects involving the metadiaphyseal portions of the first 3 metacarpals of the left hand (Fig 2); phleboliths were present. Roentgenograms of the left forearm showed thinned and slightly expanded cortices without periosteal reaction or fracture, involving the distal humerus, radius, and ulna. The metaphysis and diaphysis were unusually coarse and had a honeycombed appearance (Fig 3).

Fig 2.—Roentgenogram demonstrating cortical thinning and expansion of the metadiaphyseal portions of the first 3 metacarpals (arrows) of the left hand. A soft tissue calcification consistent with a phlebolith is adjacent to the distal phalanx of the thumb.

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Fig 3.—Plain film with 2 views of the left forearm demonstrate multiple rounded calcifications (phleboliths) scattered throughout the soft tissues. The distal humerus, the radius and ulna have thinned and slightly expanded cortices without periosteal reaction or fracture.

Magnetic Resonance Imaging

T1-weighted (TR/TE 400/14), proton density (TR/TE 1650/14) and T2-weighted (TR/TE 1650/80) images were obtained with 5-mm thick slices (Fig 4A-C). The cortical bone in the first 3 metacarpals was abnormally expanded and scalloped by a low signal intensity lesion on the T1-weighted images (Fig 4A) that increased significantly on the proton density (Fig 4B) and T2-weighted images (Fig 4C arrows). Similar abnormalities were present in the surrounding fascial planes, muscle, and subcutaneous tissue.

Additional images of the left hand and forearm were obtained using Gadolinium-DTPA (Gd-DTPA) contrast enhancement to evaluate the suspected vascular nature of the skeletal and soft tissue angiomatosis. T1-weighted (TR/TE 400/14) images with Gd-DTPA of the forearm (Fig 5) showed marked abnormal contrast enhancement throughout the extensor muscles surrounding the radius as well as within the flexor muscles anteriorly. Contrast enhancement also was present in the cortically based bone lesions. These results confirm the presence of a low-flow venous malformation infiltrating soft tissue and bone.

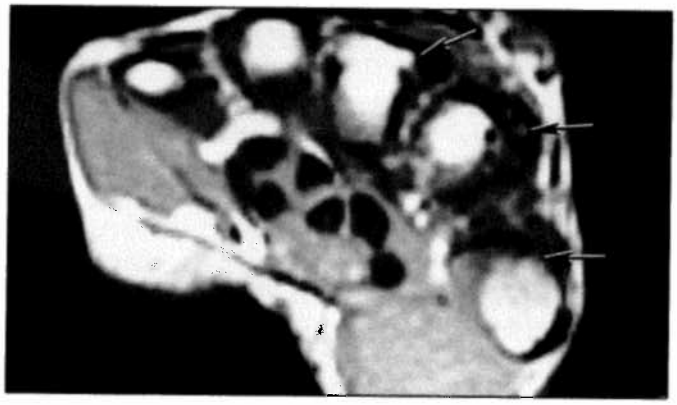

Fig 4A,B,C.—A) Magnetic resonance imaging with T1; B) proton density; C) T2 weighted sequences. The majority of the marrow fat is preserved in the metacarpals; however, abnormal low signal lesions were detected on T1 that increased in intensity on both proton density and T2-weighted images in the first 3 metacarpals (arrows). All 3 appeared to have the epicenter in the cortex with only minimal marrow involvement. The same signal abnormalities were also present in the soft tissue surrounding the affected metacarpals.

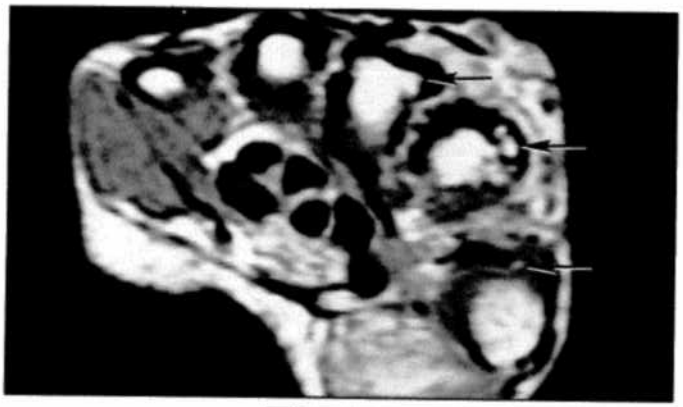

Fig 4B.—Above.
Fig 4C.—Right

Discussion

Nomenclature for vascular birthmarks is complicated by many years of confusing descriptions and classification schemes that do not correlate with clinical outcomes. For example, the word *hemangioma* has become a generic term used to describe a diverse group of vascular lesions regardless of etiology or natural history.[7] Descriptive, anatomicopathological, embryological and biological classification schemes have been proposed.[7-9] The system most commonly accepted by authors in the field today combines cellular features with clinical behavior.[7,8] There are 2 main categories: hemangioma and malformations. *Hemangioma* is limited to vascular lesions that show increased endothelial mitotic activity, a biphasic growth pattern and subsequent regression. *Malformations* are defined as vascular lesions with normal endothelial mitotic activity and fail to show regression.[9] Malformations may contain arterial, venous, capillary or lymphatic elements. The histologic term "cavernous" is confusing and has been applied to vascular lesions which may or may not involute. Mulliken[7] suggests that the term is not useful and should be avoided in the discussion of vascular anomalies.

Maffucci's syndrome is characterized by subcutaneous vascular anomalies, enchondromas and a 15% risk of developing chondrosarcomas.[9] The enchondromas most commonly involve the metacarpal and phalangeal bones, and on roentgenograms often demonstrate oval, radiolucent defects associated with expansion and cortical thinning. In this patient the detection of phleboliths suggests a low flow venous anomaly. Hand films showed small radiolucent defects and the diagnosis of Maffucci's syndrome was considered. However, roentgenograms of the forearm demonstrated coarse and honeycombed changes involving the metaphysis and diaphysis of the radius and ulna; these findings were suggestive of an intraosseous malformation. Mag-

Fig 5.—Magnetic resonance imaging of the forearm with Gadolinium-DPTA enhancement. There is marked enhancement throughout the extensor muscles surrounding the radius and penetrating the flexor muscles anteriorly. Contrast enhancement is present in the cortically based bone lesions of the ulna (U) and radius (R) consistent with a large infiltrating venous malformation.

netic resonance imaging has become an important modality in defining the extent and rate of flow in hemangiomas and arteriovenous, venous, lymphatic and lymphaticovenous malformations.[10,11] Venous malformations demonstrate slow flow on Gadolinium-DTPA enhancement (T1-weighted images only), filling defects, and enhancement on T1-and T2-weighted images.[6,7] Arteriovenous malformations show high flow rates and lymphatic malformations fail to show enhancement on magnetic resonance imaging. Lymphaticovenous malformations show focal areas of enhancement. Without evidence of a tumor mass biopsy is not required to confirm the diagnosis.[10] Thus, based on Mulliken's classification scheme, this patient has a diffuse venous malformation involving the left arm with intraosseous infiltration.

Vascular anomalies involving bone are rare; however, several specific clinical entities have been defined to include cystic angiomatosis, neonatal hemangiomatosis, Gorham's syndrome and Klippel-Trenaunay-Weber syndrome. Subcutaneous vascular malformations rarely cause radiographic changes within adjacent long bones.[12-16] Most cases presented with local pain and/or swelling. Roentgenographic findings are varied and may include: 1) Cystic osteolytic lesions, 2) "soap-bubble" or "honeycombed" medullary trabecular network, 3) expanded cortex, 4) irregular cortical thickening and peripheral sclerosis, 5) calcified mass, 6) osteolysis with soft-tissue invasion and 7) normal studies.[1,4,12-16] The differential diagnosis includes osteoid osteoma, chondrosarcoma, fibrosarcoma, malignant fibrous histiocytoma, primary lymphoma, vascular neoplasms, metastatic disease, plasmacytoma, histiocytosis X, epidermoid cysts, glomus tumors, enchondroma, lipoma, neurofibroma and osteomyelitis.[1,4,14-17]

Cystic angiomatosis of bone is characterized by osteolytic lesions (single, multiple or diffuse) with minimal cortical erosion on roentgenograms.[12,18-21] Osseous disease may be found at virtually any site; however, the most frequently reported areas are the spine, skull, pelvis, and ribs.[18] Most patients are asymptomatic and detection often is incidental with routine roentgenographic examinations.[18-22] Reid et al[18] reported a series of familial cystic angiomatosis inherited in an autosomal dominant fashion with a predilection for peripheral skeletal involvement. Pancytopenia and coagulation disorders have been associated with severe skeletal disease.[20] Some authors have reported "systemic cystic angiomatosis" involving multiple organ systems, other than bone, based on a "cystic" appearance on ultrasound or gross observation.[21] We suggest that the term cystic angiomatosis be restricted to those cases with predominantly skeletal disease; although 50% of the patients have been reported to have concomitant extraskeletal angiomas, the overriding feature is marked skeletal involvement.[18]

Neonatal angiomatosis has been divided into 2 subtypes.[9,23,24] The development of multiple cutaneous lesions with life-threatening multisystem involvement has been called diffuse or disseminated neonatal hemangiomatosis.[7,9,23,24] Children with widespread cutaneous lesions with minimal or asymptomatic visceral involvement have been classified as benign neonatal hemangiomatosis (do not require intervention).[9,23] Diffuse neonatal hemangiomatosis most commonly affects the skin, liver, lungs, gastrointestinal tract, and central nervous system.[9,24] Skeletal involvement has been reported, but is extremely rare.[24]

Gorham's syndrome, also known as phantom bone, massive osteolysis, acute spontaneous resorption of bone, disappearing bone, cryptogenic osteolysis, spontaneous absorption of bone and progressive atrophy of bone, is characterized by progressive osteolysis extending across joint spaces to involve contiguous bones.[9,14,25] More than 100 cases have been reported in the world literature, and the most frequent cause of death is respiratory complications due to lysis of the thoracic cage.[14,25] Skeletal involvement is usually monocentric and the most frequent sites of involvement are the mandible, ribs, scapula, femur, and humerus.[25] Roentgenographic changes in early stages resemble osteoporosis with intramedullary and subcortical lucency.[25] More advanced cases demonstrate tapering of the involved long bones similar to a licked stick of candy followed by complete absorption. Bone biopsies demonstrate histologically benign vascular channels with fibrosis and replacement of the normal bony architecture. Long-standing lesions develop progressive fibrosis infiltrating into adjacent soft tissue, although cutaneous angiomas have been reported at the onset.[8,25]

Klippel-Trenaunay-Weber syndrome (including Parkes-Weber variant with the addition of and arteriovenous fistulae) is characterized by cutaneous vascular lesions, venous varicosities and hypertrophy of soft tissue and bones.[9] The lower limb is most often involved in a unilateral distribution. Soft tissue and skeletal hypertrophy is believed to be secondary to increased vascular perfusion. In contrast to the hypertrophy observed in arteriovenous fistulas, patients with venous anomalies involving skeletal bones are usually normal or hypoplastic.[26] Limb hypertrophy and intraosseous involvement also has been reported with lymphatic malformations.

Detection of phleboliths on radiographs is suggestive of low flow venous anomalies and can be a significant finding in venous malformations, Maffucci's syndrome, and arteriovenous malformations.[27-28] To differentiate between these various disor-

ders additional studies are required. Arteriography, phlebography and arthrography are not recommended for children and often do not provide adequate therapeutic information.[26] Noninvasive, painless techniques for evaluation of vascular anomalies include sonography, Doppler, color Doppler, CT scans and MRI. MRI studies in patients with suspected venous malformations are superior to other studies due to the clear delineation of skin, muscle, joint and bone involvement.[26] In addition, with use of Gadolinium-DTPA, flow rates throughout the malformation can be determined.

Prognosis and therapeutic intervention is different for the various forms of vascular anomalies and vascular syndromes. In this patient it was important to exclude Maffucci's syndrome because of the increased risk of developing chondrosarcomas.[9] Individuals with arteriovenous malformations can develop pseudo-Kaposi sarcomatous skin changes, skin ulcerations, increased cardiac output and even congestive heart failure.[26] Arteriovenous malformations usually are dormant during childhood.[26] Patients who have venous malformations involving a limb complain of pain and swelling often related to vascular thrombosis.[26] Joint effusions and hemarthrosis are a consequence of vascular infiltration. Massive intraosseous disease of long bones also results in decreased bone density and increased risk of fracture, as demonstrated by this case.

Treatment of venous malformations is difficult and often unsuccessful. External support through the use of custom-made elastic stockings or wraps is indispensable and should be encouraged on a lifelong basis. If well circumscribed the patient may experience some therapeutic benefit from percutaneous sclerotherapy under fluoroscopy and surgical excision of venous pouches.[26] If knee hemarthrosis is not treated with synovectomy and surgical removal, flexion contractures, leg muscle atrophy, equinous deformity of the foot and progressive ankylosis of the joint may ensue.[26] The increased risk of fracture in this patient required us to provide him with a molded orthoplast arm guard secured with velcro straps.

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Parents' Reporting of Symptoms in Their Children: Physicians' Perceptions

➤ *(Continued from Page 217)*

Discussion

This survey was not designed to establish definitively the incidence of accurate or inaccurate reporting among parents, but rather to tap the perceptions of those who work most closely with children's illnesses. Survey results suggest that physicians are aware of the potential for both under- and over-reporting, and believe in the abstract that these form a rough normal curve. However, they do not report an equivalent number of puzzling cases in their own practices. Perhaps they are aware of those parents within their practice who falsely report/induce symptoms, or perhaps they do not believe that parents in their practice are typical. Induction of symptoms, if not persistent false reporting, is clearly child abuse. It is noted by S. Choy, Director of the Multidisciplinary Child Protective Team at Kapiolani Medical Center for Women and Children that Hawaii Child Protective Services has received fewer than 5 total reports of MBP.

The reluctance of respondents to discuss their puzzling patients is unclear. Perhaps they simply did not wish to share these cases with the author, who is not a physician, or did not feel that confidentiality could be properly safeguarded in such a discussion. Perhaps, as one indicated, they have management strategies and resources already in place. It is also possible that they are reluctant to face the implications of MBP—a personally stressful process in addition to its legal implications.[15]

It is recommended that physicians, together with other health and social services professionals, be aware of MBP. Knowledge in the abstract, however, like awareness of other forms of child abuse, is not enough. By their own estimates, approximately 1% of respondents' patients may have parents who frankly lie about